ABSTRACT OF THE INVENTION

The gene associated and causative of classical late infantile neuronal ceroid lipofuscinosis (LINCL), *CLN2*, has been identified and characterized. The translation product of this gene is a novel protease and a deficiency in this activity results in LINCL. Identification of *CLN2* will not only aid in the prevention of LINCL through genetic counseling but provides strategies and test systems for therapeutic intervention. In addition, further characterization of this previously unknown lysosomal enzyme may provide useful insights into other more common human neurodegenerative disorders. Finally, the utility of a general approach for determining the molecular bases for lysosomal disorders of unknown etiology has been demonstrated.